Johns Hopkins Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy Program

Volume 8, Issue 2 **Summer 2018**

2018 ARVD/C Family Seminar

We had a beautiful weekend for this year's seminar which was well-attended with over 150 in attendance.

Those that arrived into Baltimore early Friday evening attended a Meet 'n Greet at the Hilton Garden Inn and enjoyed some hors d'oeuvres and company of old and new friends. This was a great opportunity to interact with our staff and share stories with our attendees. This is often a favorite for seminar attendees.

On Saturday, we started off with Brittney's introductory talk on The ABC's of ARVD/C. We then heard from a number of speakers addressing topics including diagnosis, management, heart failure, ICDs, genetics, exercise, ARVC in the pediatric population, disease mechanism, catheter ablation, sympathectomy, and how to practice mindfulness in a society where we are always on the go, rarely taking the time to enjoy what life has to offer.

A number of Hopkins faculty and staff members presented including Dr. Hugh Calkins, Dr. Hari Tandri, Dr. Cynthia James, Brittney Murray, Dr. Jane Crosson, Dr. Charles Love, Dr. Nisha Gilotra, Dr. Stephen Chelko, and Dr. Neda Gould. We were delighted to have Jennifer Cory also share her work on the Heart Initiative.

In addition, research opportunities were held throughout the afternoon, including blood draws, ICD interrogations, ECGs, Holters, finger length measurements, and more blood draws. Thank you to everyone who was able to stay and participate in the various research studies. You are a vital part of our research success.

View presentations now at:
http://tinyurl.com/2018ARVDSeminar

SAVE THE DATE – Our 20th Seminar Anniversary
➔ Saturday, May 4th, 2019 ←

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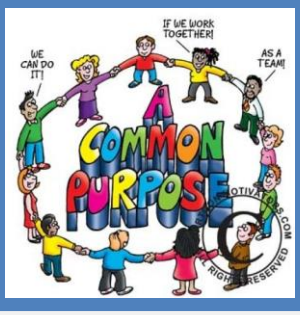

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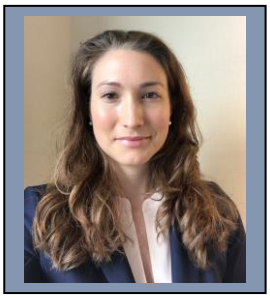
Meet Our ARVD/C Research Program Team Members

Dr. Apurva Sharma is a graduate and post graduate in Internal Medicine from one of the premier institutes of India, King George's Medical University. Dr. Sharma brings with her a wealth of clinical exposure from India which was further sharpened with a combination of research training and electives at Mt. Sinai, St Luke's Roosevelt Hospital, New York and a residency in Internal Medicine at Saint Peter's University Hospital/Rutgers Robert Wood Johnson Medical School, New Jersey. Through the years, she was the recipient of research awards and has authored numerous articles and presentations in the field of Cardiology. Her research interests include a strong emphasis on translational and innovative patient-oriented research with special interests in cardiomyopathy and preventive cardiology. She has joined the Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) group as a post-doctoral fellow this July and would be leading the project on assessment of risk of family members with ARVC.

Weijia Wang, MD attended Zhejiang University in China for his medical education between 2005 to 2012. He finished his internal medicine residency at Tufts Medical Center in Boston and became board certified in 2016. He acquired a strong interest in clinical research and earned a Master of Public Health degree with a concentration in Epidemiology and Biostatistics from the Johns Hopkins Bloomberg School of Public Health in May 2017. Weijia was awarded the Clinical Research Award in Honor of Mark Josephson and Hein Wellens from the Heart Rhythm Society for his work on how exercise restriction affects disease outcomes in ARVD/C. He continued working with the ARVD/C group as a research fellow, making great contributions to our understanding of exercise, as well as creating useful database systems. He is now a cardiology fellow at University of Massachusetts Medical School and we wish him the best.



Julia Cadrin Tourigny MD, completed her medical education and training in Cardiology at the Université de Montréal and was certified by the Royal College of Physicians of Canada (FRCPC) in 2015. Following her strong interest in clinical research, she obtained a Master of Biomedical Sciences for her work on the efficacy of amiodarone in patients with atrial fibrillation and heart failure. She then pursued additional training in cardiac electrophysiology at the Montreal Heart Institute from 2015 to 2017. Over the past year and a half, she worked as a postdoctoral clinical and research fellow in inherited heart disease at the Johns Hopkins Hospital under the George Mines Traveling Fellowship from the Canadian Heart Rhythm Society. During her time with us, she led an important multicenter project aiming to improve the arrhythmic risk stratification in patients with ARVC. We are looking forward to this collaborative project being published soon and wish her all the best as she returns to Montreal this summer.

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The Johns Hopkins Arrhythmogenic Right Ventricular Cardiomyopathy and Complex Arrhythmia Precision Center of Excellence

The Johns Hopkins ARVD/C Program has been chosen as one of eight centers to launch a "precision medicine center of excellence to highlight areas where the newest technologies and measurement tools can be applied to greatly improve patient care." Proposed goals include 1) expanding access to multidisciplinary expert care, 2) optimizing early detection through genomic, imaging, and epidemiological approaches, and 3) developing patient-specific therapies for those at highest risk of sudden cardiac death and/or heart failure.

We are excited to continue our important work in understanding the underlying disease mechanism so that we can develop new and better treatments for each of you. We will continue to expand both our clinical and research avenues to achieve these goals. Thank you for your continued support.

Please visit our new website showcasing this important work:
https://www.hopkinsmedicine.org/inhealth/precision-medicine-centers/arvc

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Clinical Services at Johns Hopkins

The Johns Hopkins ARVD/C Program provides a variety of clinical services. We see patients for second opinion consultations to discuss diagnosis and management, genetic counseling and testing, routine ICD management and family member screening. We can also arrange concurrent cardiac testing.

Patients are seen in consultation with Dr. Hugh Calkins or Dr. Hari Tandri and our clinical genetic counselor, Brittney Murray, to discuss test results, family history, and to provide guidance regarding further management. We see all of our patients for genetic counseling to discuss the diagnosis, the psychosocial impact of living with ARVD/C and with an ICD, as well as to discuss the benefits and limitations of appropriate genetic testing. In selected cases we also offer catheter ablation as a treatment for difficult to manage ventricular tachycardia. Appointments with our heart failure specialist, Dr. Nisha Gilotra can also be arranged. These appointments are billed to your health insurance. To schedule an appointment, contact Crystal.

Tele – Genetic Counseling

We continue to take steps to expand our clinical services via telemedicine for those unable to travel to Baltimore. This service currently consists of the genetic counseling/genetic testing appointment only. We hope to be able to offer second opinion consultations with our physicians in the future. Telemedicine appointments are currently not billable to your insurance company and require payment out of pocket. This service may not be available in certain states. To discuss your eligibility or to schedule an appointment, contact Crystal.

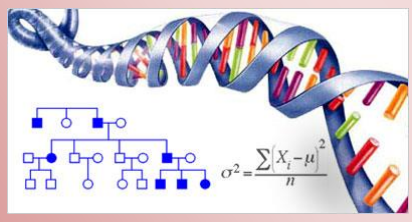
Crystal Tichnell, MGC ctichnell@jhmi.edu 410-502-7161

What's New?

Direct-To-Consumer Genetic Testing

Direct-to-consumer (DTC) genetic testing has become quite popular as consumers attempt to learn about their health risks, common traits, and ancestry. It is important to understand that DTC genetic testing is not meant to be a substitute for ARVC genetic testing, or any clinical genetic test for that matter. In fact, studies have reported that a significant number of genetic variations identified through DTC testing have been identified as false positives. We recommend that you discuss genetic testing options with your genetic counselor and physician so that you understand the type of results you can expect. Read a brief article below written by Brittney and our genetic counseling colleagues:

https://www.nature.com/articles/s41436-018-0097-2



Flecainide Trial

The Johns Hopkins ARVC program will be participating in a pilot study focusing on the antiarrhythmic effects of flecainide and its safety among ARVC patients. Medication research within the ARVC population has been difficult given that our patients tend to be on various "cocktails" to find that right combination to manage your arrhythmias. This will be an opportunity to solely look at one medication for treatment of arrhythmias and assess its efficacy and safety. This is an exciting opportunity and will pave the way for additional clinical trials. Stay tuned for more information regarding your eligibility to participate.

Exercise

Exercise remains an important topic when discussing management recommendations of patients diagnosed with ARVC or who carry a genetic mutation associated with ARVC. How much exercise is ok? At what intensity? What types of exercise? We continue to invite registry enrollees to participate in a lifetime exercise interview via telephone, so be on the lookout for those email invitations from Julia! Thank you for your willingness to participate.

Tips for Managing Your HealthCare

1. Keep a list of your current medications
2. Keep a list of medications you have tried in the past to manage your arrhythmias
3. Keep all of your healthcare providers up-to-date. Make sure everyone is receiving copies of your medical records. This does not always happen automatically.
4. Keep a copy of your medical records for yourself. This way you can fill in the missing pieces.
5. Make sure your device is checked on a regular basis. It is important to have an in-person check at least once per year to ensure your device receives important updates.
6. If you have a device that is compatible with home monitoring, SET IT UP! This can save you an in-person visit once or twice a year and can come in handy if you're not feeling quite right.
7. Don't wait until you are out of medication to request a refill. Our office is not always available to submit medication refill requests urgently. Refill requests received via fax from your pharmacy work best (Fax: 443-873-5073)
8. We will complete medication refill requests as long as you are followed by our office at least once every 2 years.
9. Schedule your appointments at the recommended intervals. This helps us to keep a close eye on progression and make necessary adjustments to your treatment plan.
10. Maintain contact with your genetic counselor as genetic variations are reclassified from time to time.

Ongoing Research Opportunities at Johns Hopkins

Relative Safety, Efficacy, and Patient Satisfaction of Standard ICDs versus the Sub-Cutaneous ICD (S-ICD)

Who can participate? Patients diagnosed with ARVD/C and have:

- ✓ S-ICD implanted
- ✓ Transvenous ICD implanted after January 2013

What do I have to do? Contact Crystal (ctichnell@jhmi.edu). You will need to sign a consent form, send us your medical records, and complete online questionnaires.

If you have an S-ICD and have not been in touch with Crystal, please email ctichnell@jhmi.edu to enroll in this important study.

Do I have to travel to Johns Hopkins? No

Once enrolled, please remember to check your email for reminders to complete the online questionnaires. It is really important that we continue to collect this follow up data. Thank you for your participation!

Email Crystal to discuss your eligibility and enrollment.

Sponsored by Boston Scientific
PI: Hugh Calkins, MD
Johns Hopkins IRB NA_00042471 *(Predictors of Implantable Cardioverter Defibrillator (ICD) Firing in Right Ventricular Dysplasia)*

Clinical and Genetic Investigations of Right Ventricular Dysplasia (ARVD/C Registry)

Who: Children and Adults with ARVD/C

What: Collection of pertinent past medical records and continued collection for 5 years. A blood sample for DNA for genetic mapping of ARVD/C genes will also be collected.

How to Join: Contact Crystal at 410.502.7161 or ctichnell@jhmi.edu.

Have you had an epicardial ablation?

We are looking for people with ARVD/C who have had an epicardial ablation to join our Registry. Help us discover how this new technique affects the course of ARVD/C! Contact Crystal at 410-502-7161 or ctichnell@jhmi.edu

Outcomes of genetic counseling for arrhythmogenic cardiomyopathy: A comparison of face-to-face and tele-genetic counseling

As we've mentioned before, we are now offering tele-genetic counseling! We are taking this opportunity to determine whether genetic counseling services offered via telephone/ videoconference differs from the traditional face-to-face genetic counseling in achieving three key cardiac genetic counseling outcomes: reducing cardiac-specific anxiety, increasing disease-specific genetic understanding, and enhancing patient empowerment. Data will be collected by completing questionnaires 2 weeks preceding and 2 weeks following a genetic counseling session for an arrhythmogenic cardiomyopathy indication. Results of the study will provide some of the first evidence of genetic counseling outcomes in cardiology clinics, and also contribute outcome data of alternative methods to expand genetic counseling services. **THANK YOU TO EVERYONE WHO HAS PARTICIPATED IN THIS STUDY!!!** We will be pausing data collection for the summer and will begin data analysis. **STAY TUNED!**

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Heart Rhythm Society Abstract Presentations

The 37th Annual Heart Rhythm's Scientific Sessions were held in Boston in May 2018. The research and collaborative efforts of the Johns Hopkins ARVD/C Program were once again well-represented by several presentations by our team and collaborators. Below you will find a summary of some of the abstracts presented at the meeting.

INDIVIDUALIZED ARRHYTHMIC RISK PREDICTION IN A PRIMARY PREVENTION ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY (ARVC) POPULATION: A TRANSATLANTIC MULTINATIONAL COLLABORATION

Julia Cadrin-Tourigny, et al.

The purpose of this study was to design a model that can predict your risk of developing ventricular arrhythmias. The study included 976 individuals meeting the diagnostic task force criteria without prior history of sustained ventricular arrhythmias from five different registries throughout North America and Europe. The clinical predictors utilized in the model to determine the annual risk of a sustained ventricular arrhythmic event include: male gender, age, recent syncope, prior non-sustained ventricular tachycardia, 24 hour PVC count, number of leads with T-wave inversions, and right ventricular ejection fraction. A paper with additional details has been submitted for review.

ARRHYTHMIC COURSE OF ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY PATIENTS WITHOUT IMPLANTABLE DEFIBRILLATORS

Weijia Wang, Julia Cadrin Tourigny, Aditya Bhonsale, Crystal Tichnell, Brittney Murray, Cynthia James, Harikrishna Tandri, and Hugh Calkins.

The goal of this study was to report the arrhythmic course of ARVC patients without an ICD or who had delayed implant (more than 6 months after diagnosis). The study population consisted of 131 patients meeting the above mentioned criteria. At the time of diagnosis, ICDs were not recommended to 58 (44%) patients and declined by 22 (17%). An additional 40 (31%) patients who actually met the task force diagnostic criteria were not recognized as having ARVC by their treating physician. Over the course of 8 years, 38 (29%) patients experienced an arrhythmic event (8 cardiac arrests and 30 sustained ventricular arrhythmias) while not having an ICD. Spontanenous sustained ventricular arrhythmias, syncope, male gender, proband status, competitive athlete, and inducibility in electrophysiology study predicted arrhythmic events.

DOES SIZE MATTER? PANEL SELECTION IN GENETIC TESTING FOR ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY (ARVC)

Brittney Murray, Crystal Tichnell, Hugh Calkins, Daniel Judge, and Cynthia James

The purpose of this study was to determine if utilizing a large panel would increase detection of pathogenic variants among those undergoing genetic testing for ARVC. The small genetic testing panels consisting of the desmosomal genes (PKP2, DSP, DSC2, DSG2, JUP) are able to identify a pathogenic variant in 50-60% of cases. With overlap between genetic cardiac conditions, larger panels consisting of 60-100+ genes are now available. Of 162, 78 (48%) had an identified pathogenic variant in the 5 desmosomal genes. However, 10 (6%) had large deletions/duplications only detected on next-generation panels. In addition, 13 (9%) had predicted pathogenic variants in other genes (PLN=5, LMNA=2, SCN5A=1, MYBPC3=1, RBM20=1, MYH7=1, and an incidental KCNQ1 variant. Utilizing larger gene panels significantly increased sensitivity from 50-60% to 63%. Furthermore, large gene panels can identify a small group of patients that have digenic (2 genetic changes) inheritance.

Your Support Makes Breakthroughs Possible

As a charitable, tax-exempt organization, Johns Hopkins Medicine relies on donations to make a difference in the lives of our patients. Supporters of Dr. Calkins, Dr. Tandri, and their team of experts in the ARVD/C Program, become part of our mission to provide exceptional personalized care and to find better ways to diagnose and treat our patients. Here are some of the many ways that you can help:

Make a Donation

Donations of all sizes, whether they're one-time or recurring, make a difference and can be made online at **www.arvd.com**, by phone at **443-287-7382**, or by mail (information listed below). There are a variety of ways to make a gift to support our efforts in the ARVD/C Program:

- Make an outright gift of cash or securities
- Become a monthly donor
- Give in honor or in memory of a loved one
- Give through IRA's, wills and trusts
- Leverage matching gifts through your workplace

Fundraising

There are many opportunities to become personally involved in raising awareness and much-needed funds on behalf of the Johns Hopkins ARVD/C Program:

- Create an online giving page and leverage social media
- Ask friends to make contributions in lieu of gifts
- Host your own event or auction
- Plan a fundraising event in your community or school
- Contribute a portion of your company's sales

The Johns Hopkins Heart and Vascular Institute Development Office is here to help!

We welcome your questions, concerns, ideas, and feedback. Please contact **Adrienne Rose,** Senior Associate Director of Development, at 443-287-7382 or arose25@jhmi.edu, for more information.

Gifts by Mail:

The Johns Hopkins Heart and Vascular Institute
600 North Wolfe Street, Blalock 536B
Baltimore, MD 21287
***Indicate the "ARVD/C Program" on the memo line**

ARVD/C Program Info

ARVC Program Staff

Hugh Calkins, MD—Director
Harikrishna Tandri, MD—Faculty
Cynthia James, ScM, PhD—Genetic Counselor
Brittney Murray, MS—Genetic Counselor
Crystal Tichnell, MGC, RN—Genetic Counselor
Anna Burton—Genetic Counselor Assistant
Giovanna Jimenez—Clinic Coordinator
Julia Agafonova—Research Assistant
Apurva Sharma—Research Fellow

Don't forget to keep us informed of your most up-to-date contact info! Please send any changes and updated medical records to Crystal at ctichnell@jhmi.edu Thank you!

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600 North Wolfe Street; Blalock 545
Baltimore, Maryland 21287

P: 410-502-7161 / F: 443-873-5073

CONTACT US

www.ARVD.com

Support Group Info

Looking for a support group?

FACEBOOK Groups (private):
- ARVD/C Youth Society
- Hope for ARVD
- The Broken Heart Club – ARVD Edition –
- ARVC can't stop me from…

Precision Medicine WebPage:
Read "Feeling the Beat": Stories from the ARVC Community here
https://www.hopkinsmedicine.org/inhealth/precision-medicine-centers/arvc

ARVD/C Mentor Program:
Get matched with an ARVD/C mentor!
Connect with a mentor who has navigated the challenges of life with ARVD/C and receive:

- Support
- Connection
- Understanding
- Strategies for Thriving with ARVD/C.

Contact Nancy Bogle at nbstjohn@gmail.com for more information and visit ARVDHEARTANDSOUL.org

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